GEUVADIS-Sequencing Survey - Final

1/11 - Introduction

THE GEUVADIS PROJECT - More information on our website: www.geuvadis.eu
The latest high-throughput next-generation sequencing technologies allow investigators to sequence entire human genomes and transcriptomes at an affordable price and within a short time frame. An increasing number of research centers in Europe have access to these technologies, in-house or through regional, national and international infrastructures. Storing, disseminating and analysing the large amount of data produced generate major challenges. Tackling these challenges requires extensive exchange of data, information and knowledge between sequencing centers, bio-informatics networks, the medical research community and the industry at the European level. The GEUVADIS (Genetic EUropean VAriation in DISease) Consortium has four main aims:

- Develop standards in quality control and assessment of sequence data
- Develop models for sequencing data storage, access and exchange
- Develop standards for the handling, analysis and interpretation of sequencing data from DNA (and RNA)
- Develop guidelines on the handling of ethical, legal and social implications of phenotype prediction from sequence variation

PURPOSE OF SURVEY

- Collect quantitative and qualitative information on the current status of DNA sequence production, storage, exchange and analysis in Europe.
- Collect feedback from research/clinical centers on their main challenges and difficulties regarding the management of these large data-sets potentially containing important medical information
- Collect information on local standardization efforts, and avoid duplication of efforts throughout Europe
- Create a road-map/policy document outlining the necessary steps to take national standards to the European level

POTENTIAL PUBLICATION OF RESULTS
Results of this survey will be presented at an internal GEUVADIS workshop, on October 30-31st 2012.
We will disseminate the road-map to potential funders (private of public), including the European Commission and other possible national public funders. We will submit an abstract to the Joint Conference of Human Genome Meeting 2013 and 21st International Congress of Genetics.

DEADLINE EXTENDED: 10.10.2012

Instructions to fill-in the survey
The survey should not take you more than 15 mins.
It is structured as follows:

1. Introduction
2. Confidentiality agreement
3. Contact details
4. Main characteristics of the Institution
5. Technology (number/type of sequencers, nb of runs, capture method)
6. Data Use (clinical/research uses)
7. Patients/participants and you (origin of samples, consent forms, incidental findings...)
8. Data storage
9. Data analysis (general information on analysis pipelines)
10. Clinical data management
11. Problems and suggestions

If possible, make sure you have all the information at hand to fill-in the entire survey. If you cannot finish the survey in one shot, your answers will be saved on your computer, and on your Internet browser for 2 weeks. To come back to the survey later, make sure you use the same computer and the same browser. Make sure you use the back and forth buttons from Qualtrics and not from your browser to ensure that all your answers are saved.

I have read and understood the instructions
❍ Yes

2/11 - Confidentiality Agreement
Please enter your preferences below
- ❑ I want my name and personal information to be kept confidential
- ❑ I want the name of my Institution to be kept confidential
- ❑ I want to be informed of all future publications of the results

3/11 - Contact Details
Q3.1 First name
Q3.2 Last Name
Q3.3 Email address

4/11 - Main characteristics of the Institution
Q4.1 Your Institution is:
- ❑ Research Center
- ❑ Clinical Center
- ❑ Hospital Department
- ❑ University
- ❑ Private Company

Q4.2 Institution Name
Q4.3 City
Q4.4 Country

Q4.5 Main source of funding for your institution
- ❍ Public funding
- ❍ Private funding

THIS DOCUMENT IS A PREVIEW OF THE GEUVADIS SURVEY

TO TAKE THE SURVEY PLEASE VISIT: https://qtrial.qualtrics.com/SE/?SID=SV_bHJDue550PwdFrv

ONLY COMPLETED ONLINE SURVEYS WILL BE TAKEN INTO ACCOUNT

FOR ANY QUESTION CONTACT Gabrielle Bertier

5/11 - Technology

Q5.1 Please indicate if the sequencing is produced:
- ❑ Locally - In-house facilities
- ❑ Externally

Q5.2 If 'Externally', please indicate if the sequencing data is produced:
- ❍ Data produced by a company
- ❍ Data produced by another center

Q5.3 Specify name of the center/company

Q5.4 Technology available in house

	Illumina MiSeq	Illumina GAII	Illumina HiScanSQ	Illumina HiSeq1000	Illumina HiSeq2000	SOLiD 4	SOLiD 5500	SOLiD 5500xl	Roche GS Junior	Roche 454FLX+	Ion Torrent Ion PGM	Ion Torrent Ion Proton	Other
Number of machines													
Average runs/year													

If Other; specify which machine you are using

Q5.5 Targeted Resequencing: please indicate the enrichment method(s) used in your institution
- ❑ Long-PCR
- ❑ Access Array System (Fluidigm)
- ❑ Microdroplet PCR (Raindance)
- ❑ AmpliSeq technology (Life Technologies)
- ❑ TrueSeq Amplicon Kit (Illumina)
- ❑ HaloPlex (Agilent)
- ❑ SureSelect (Agilent)
- ❑ SeqCap EZ (Nimblegen)
- ❑ TrueSeq Enrichment Kit (Illumina)
- ❑ TargetSeq (Life Technologies)
- ❑ Other

Q5.6 Please specify which other capture method you are using

6/11 - Data use

Q6.1 Please indicate if the NGS data in your institution is used for :
- ❑ Research purposes
- ❑ Clinical/Diagnostic purposes

Q6.2 Type of data used for Research
- ❑ Whole genome sequencing
- ❑ Whole exome sequencing
- ❑ Targeted resequencing (custom panels)

Q6.3 Type of data used for Clinical/Diagnostic purposes
- ❑ Whole genome sequencing
- ❑ Whole exome sequencing
- ❑ Targeted resequencing (custom panels)

7/11- Research NGS: Participants and you

Q7.1 Origin of samples for research: Please indicate if the samples used for research in your institution are:
- ❑ samples from a Hospital/Clinic
- ❑ samples from other research projects
- ❑ specific collection of samples from research participants
- ❑ local biobank
- ❑ international biobank

Q7.2 Please specify from which biobank the samples for research are from:

Q7.3 Consent form: Do you have a consent form template for your NGS research activities ?
- ❍ Yes
- ❍ No

Q7.4 Consent includes right to withdraw from the research
- ❍ Yes
- ❍ No

Q7.5 Consent includes possibility to use samples/data for future research
- ❍ Yes
- ❍ Only if specifically agreed by the patient
- ❍ No

Q7.6 Open consent, for a variety of unspecific research projects
- ❍ Yes
- ❍ No

Q7.7 Incidental medical findings Please indicate if incidental medical findings from your research activities are:
- ❍ never looked at: data is filtered out to avoid this possibility
- ❍ never reported
- ❍ possibly reported if chosen by the patient
- ❍ always reported

Please describe here how you filter out your NGS data to avoid medical incidental findings:

Q7.9 If only a set of diseases are reported, please indicate below which diseases:

Q7.8 If reported, how is the medical relevance evaluated?
- ❑ on a case per case basis
- ❑ • by the Investigator
- ❑ • by thpatients's medical doctor
- ❑ • by a committee including investigators and medical doctors
- ❑ only a set of specific diseases are reported

7/11 - Clinical NGS: Patients and you

Q7.10 Do you have a consent form template for your NGS clinical/diagnostic activities ?
- ❍ Yes
- ❍ No

Q7.11 Main items of the consent form:
- ❑ Consent includes possibility to use the samples for research
- ❑ Consent includes possibility to use the data for research
- ❑ Consent includes possibility to use the samples and data for research

Q7.12 Incidental findings (not on the disease in question): Please indicate if medical incidental findings on characteristics that were not looked for are:
- ❍ never looked at: data is filtered out to avoid this possibility
- ❍ never reported
- ❍ possibly reported if chosen by the patient
- ❍ always reported

Q7.13 If reported, how is the medical relevance evaluated?
- ❑ on a case per case basis
- ❑ • by the Investigator
- ❑ • by thpatients's medical doctor
- ❑ • by a committee including investigators and medical doctors
- ❑ only a set of specific diseases are reported

Q7.14 Please describe here how you filter out your NGS data to avoid medical incidental findings:

Q7.15 If only a set of specific diseases are reported, please indicate below which diseases:

8/11 - Data storage

Q8.1 Raw data storage
- ❍ Local database
- ❍ Shared database
- ❍ Cloud

Q8.2 Please indicate if the local database for raw data storage is located in:
- ❍ Lab
- ❍ Hospital
- ❍ Other, please specify _____

Q8.3 How is the database shared?
- ❍ by research project
- ❍ with local investigators
- ❍ with local and international collaborators

9/11 - Data Analysis

Q9.1 Please indicate if the NGS data is processed in your institution by:
- ❍ Local cluster
- ❍ Supercomputing center

Q9.2 Name of the Supercomputing center where data is processed:

Q9.3 Do you have an agreement with local center, or company for raw NGS data processing?
❍ Yes
❍ No

Q9.4 Please specify parties and nature of the agreement

Q9.5 Analysis pipeline(s) Please indicate the type of pipeline(s) you are using
❑ Pipeline designed locally
❑ Pipeline designed by the sequencer's company
❑ Using a publically available or shared analysis pipeline
❑ Data processed by a specialized company

Q9.6 Please specify which pipeline you're using:

Q9.7 Specify which company processes your data:

10/11 - Patient clinical data management

Q10.1 Source of clinical information
❑ from the patient/participant
❑ from the patient MD
❑ other, please specify _____

Q10.3 Reporting of clinical results: Please indicate how NGS clinical results are reported to the patients
❑ report directly sent to the patient
❑ report sent to MD
❑ other, please specify _____

11/11 - Problems and suggestions

Q11.1 Please tell us more about your sequencing experience here, focusing on main challenges and main suggestions to overcome these challenges: Data production (Standardization of protocols, kits versions management, cost of sequencing)

Q11.2 Data storage (Storage capacity, data security, data access policy...)

Q11.3 Data exchange (Definition of access policy, protection of confidentiality of clinical and research findings)

Q11.4 Data analysis (Pipeline design, bio-informatics capacity...)